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Aortic regurgitation in newborn

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SUMMARY Clinical signs of aortic regurgitation were noted after birth in an infant who died suddenly at the age of 18 months. Congenital malformation of the aortic root with an aneurysm of the right aortic sinus and disconnection of the corresponding cusp were found at necropsy.

Aortic regurgitation occurs in infancy or later usually as the result of congential malformations of the aortic root or rheumatic disease of the aortic cusps. We have recently observed an infant who died at age 18 months with a congenital malformation of the aortic root presenting as aortic regurgitation from birth. The case reported here appears to be unique.

Case report

The patient was a full-term female baby, birth weight 3750 g. Pregnancy and family history were uneventful. Physical examination soon after birth showed no abnormality except those of the cardiovascular system. No features of Marfan's or other connective tissue disorders were present. Auscultation revealed a to-and-fro murmur, grade 4/6, with maximal intensity along the left sternal edge; phonocardiography showed that the murmur consisted of a systolic ejection and an early diastolic regurgitant component. Brachial and femoral pulses were of bounding quality.

Electrocardiogram showed sinus tachycardia and severe left ventricular hypertrophy.

Chest *x*-ray film disclosed pronounced enlargement of the cardiac shadow.

Cardiac catheterisation and angiography under general anaesthesia at the age of 1 month showed that pressures in the right atrium, left atrium, right ventricle, pulmonary artery, and left ventricle (entered through a patent foramen ovale) were, respectively (-1), (4), 25/0, 18/7, 78/0/8 mmHg. There was no difference in oxygen saturation between the right atrium and the pulmonary artery. Arterial saturation was normal.

Left ventriculography showed moderate enlargement of the left ventricular cavity with considerable thickening of the left ventricular walls. The aorta was dextroposed and its ascending portion and arch much enlarged. At the level of the aortic ring an aneurysm the size of a nut was observed and was thought to originate from the right aortic sinus (Fig. 1). Coronary arteries were thin; pseudocoarctation of the aortic isthmus was suspected.

The infant was digitalised and subsequent clinical course was characterised by slow weight

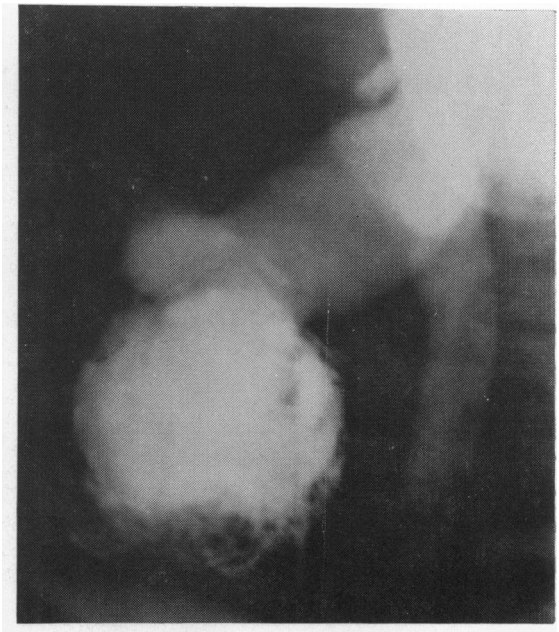

Fig. 1 *Left ventriculography lateral view.*

gain (8 kg before death), difficulties in feeding, and tachycardia. Cardiovascular findings remained unchanged throughout her life. Recatheterisation and surgical repair of the defect were planned but the patient died suddenly at the age of 1½ years.

NECROPSY FINDINGS

The heart weighed 110 g. Cardiomegaly was the result principally of hypertrophy of the left ventricular wall. The right ventricle was hypoplastic. The pericardial surfaces of the heart were normal. The left anterior and the posterior non-coronary aortic cusps were normal, while the right anterior cusp was not connected for one-half of its base to the aortic ring (Fig. 2). The corresponding aortic Valsalva sinus was aneurysmal. The aneurysm was 10×7 mm, not ruptured, and protruded into the right ventricular cavity. There was no suggestion of recent or old bacterial endocarditis.

Histological examination of aortic sinus aneurysm disclosed the presence of connective collagen tissue without evidence of inflammation. The aortic wall showed no signs of cystic medial necrosis and several myocardial sections showed only distinct hypertrophy of muscular fibres.

Discussion

Congenital malformation of the aortic root usually consists of aneurysm of the Valsalva sinuses while semilunar cusps may be fenestrated or bicuspid. Clinical manifestations ensue in infancy or later when the malformed structures either rupture or became fibrotic and calcific. In the present case a congenitally weak junction of the heart and the aorta, which is usually responsible for aneurysm of the aortic sinus, probably led to failure of connection of the right aortic cusp to the aortic ring (Fig. 2). The latter was in turn responsible for the aortic regurgitation present at birth, as the aneurysm of the corresponding aortic sinus was found to be intact both at angiography (Fig. 1) and at necropsy. The cause of the fetal malformation remains unknown.

Clinically, aortic regurgitation should be considered in the differential diagnosis of a bounding pulse and to-and-fro murmur in an infant.

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Fig. 2 *Necropsy specimen showing a probe passing through the base of the right aortic cusp into the ascending aorta. The aneurysm of the aortic cusp is located just behind and below that portion of the aortic ring which has no connection with the cusp.*